

## Dysmorphology Meeting Audit June to December 2004

Some of you may be aware that we have audited three Dysmorphology meetings, June, September and December 2004. Here is a summary of our findings –it makes riveting reading!!.

212 cases were presented in total (June = 51, Sept =75, Dec = 86). The average time taken per case was 3.6 minutes but varied from 1.5 minutes to 12 minutes (but that was a whole family!)

82 were presented with known diagnoses and the audience agreed with 79 of these . However, many of the known diagnoses had been confirmed by a specific test, 32 by chromosomes or FISH (18 of these were telomeric abnormalities) and 16 by mutation analysis. 34 (41%) were made on clinical grounds and 4 of these had suggestive radiological features.

Some cases were presented with a probable or likely diagnosis (n=58). 29 (50%) of these were confirmed as the correct diagnosis by the audience, 16 (27.5%) were refuted and for 13 (22.5%) cases we sat on the fence!

The table below shows that we were much better at making diagnoses in the morning, than the afternoon and the least useful time was the last session:

### Suggestions made for unknown cases

Time	Diagnosis			
	Definite	V. likely	possible	No suggestions
Morning	0	8	7	5
After coffee	0	9	6	7
After lunch	1	3	9	7
After tea	0	1	5	10

Finally, we looked at usefulness scores. This, I admit is highly subjective! A case was considered 'useful' if it fulfilled one of the following:

Teaching case (known chromosome abnormality or mutation) (n=48)

If the clinical diagnosis was agreed with and thereby confirmed (n=63)

If the clinical diagnosis was refuted (n=16)

For unknown cases, if a definite or very probable diagnosis was suggested (n=22)

This means that about 70% of the cases were useful!

In conclusion it is obviously advisable to rotate the presentation times for each centre, which has already been introduced and perhaps do a little less sitting on the fence!

Sahar Mansoor

Helen Stewart

Jill Clayton-Smith